
FRIEDREICH ATAXIA

Friedreich ataxia (FRDA, also called FA) is a neuromuscular disease that mainly affects the nervous system and the heart.

FRDA affects about 1 in 50,000 people worldwide, making it the most common in a group of related disorders called **hereditary ataxias**. It shouldn't be confused with a group of diseases known as autosomal dominant spinocerebellar ataxias.

FRDA is a genetic disorder with **autosomal recessive inheritance**, caused by a mutation in the frataxin gene. Autosomal refers to the fact that the frataxin gene is on chromosome 9, one of the 22 pairs of autosomes (chromosomes other than the X or Y). Recessive means it takes two copies of the disease-causing gene mutation — one inherited from each parent — to cause FRDA. People with only one copy of the gene mutation do not have any symptoms and may not know they are carriers of the gene.

About **1 in 100 Americans is an FRDA carrier**, but in some ethnic groups the frequency is higher. For example, about 1 in 70 people of Acadian (Cajun) ancestry is a carrier. In general, children with a biological sibling affected by FRDA have a 25% chance of developing the disease themselves.

Your MDA Care Center team or **genetic counselor** can give you more information about the risks of inheriting or passing on FRDA.

Symptoms of FRDA usually start **between 10 and 15 years of age**, but FRDA has been diagnosed in people from ages 2 to 50 years old. FRDA is progressive, meaning it gets worse over time, but the sequence and severity of progression varies.

Loss of reflexes occurs in most people with FRDA. Genetic testing for frataxin mutations is highly reliable and can be used to confirm or exclude a diagnosis of FRDA in almost all cases. At some point, specialized tests may be ordered to evaluate the function of muscles and nerves.

A neurologist will use **several tests to reach a diagnosis** of FRDA. Typically, diagnosis begins with a basic physical exam and a careful assessment of personal and family history. During the physical exam, the neurologist is likely to devote time and attention to testing reflexes, including the knee-jerk reflex.

Although there is no cure for FRDA yet, treatments exist for cardiac symptoms, and there are ways to manage impaired coordination, muscle weakness, and other complications of FRDA.

Many people with FRDA lead active lives, going to college, holding careers, getting married, and starting families.



WHAT SHOULD I KNOW ABOUT FRDA?

- 1.** FRDA is an inherited progressive disease that affects men and women equally.
- 2.** FRDA has an autosomal recessive inheritance pattern, requiring one copy of the disease-causing gene mutation from each parent.
- 3.** There is a wide range in age of onset and rate of progression. Symptoms can start in early childhood through late adulthood, although symptoms most often are seen in the early teens.
- 4.** FRDA first affects the legs and torso, causing unsteady walking, frequent tripping, and poor performance in sports. Balance and coordination continue to decline over time, and muscles in the legs become weak and easily fatigued, making it increasingly difficult to walk. Later, people with FRDA may have difficulty with speech, known as dysarthria.
- 5.** FRDA affects the function of the cerebellum, the part of the brain that helps plan and coordinate movements. FRDA does not affect the parts of the brain involved in mental functions.
- 6.** Individuals living with FRDA may also experience sensory impairment, skeletal abnormalities, cardiac abnormalities, and/or diabetes.

HOW IS FRDA TREATED?

In February 2023, the US Food and Drug Administration (FDA) granted approval to **omaveloxolone (SKYCLARYS™)** for the treatment of FRDA in adolescents and adults ages 16 and older. To date, SKYCLARYS is the first disease-modifying drug approved to treat FRDA. SKYCLARYS will be made available in the United States and marketed by Reata Pharmaceuticals. For more information on SKYCLARYS™, read this MDA Quest blog post: **FDA Approves Reata's SKYCLARYS for Treatment of FRDA.**

Physical, occupational or speech therapy can help with managing symptoms and improve function. People with FRDA should practice regular exercise, including appropriate balance, coordination, and core exercises. For some people with FRDA, muscle conditioning, strengthening, and stretching may be helpful. Speech therapy can help when speech clarity is affected, especially in younger patients. Some patients may decide to use speech-generating devices for additional support.

About 11% to 15% of people with FRDA experience painful muscle spasms or contractions. These symptoms may be managed with physical therapy, as well as medications such as **muscle relaxers and pain relievers.**

People with FRDA may develop difficulty understanding speech. This can be due to hearing loss or an impaired ability to process sounds after they reach the ears. **Evaluation by an audiologist** with specific testing for hearing loss and auditory processing disorder will provide the best treatment.

Vision loss can occur later in the disease. If vision loss occurs, **modifications for people with visual impairment** can be helpful, such as using large-print materials, magnifying devices, and so forth. If an individual is experiencing vision loss, they should be evaluated by an ophthalmologist.

People with FRDA can develop dysfunction of the beta cells in the pancreas that normally produce insulin, and 10% of them develop diabetes. In addition, 20% of people with FRDA have a condition called carbohydrate intolerance, which is an inability to effectively process carbohydrates — sugars and starches — into energy. Patients who develop glucose intolerance or diabetes may be required to **follow an insulin regimen** and/or consult with a dietitian to modify their diet.

Heart abnormalities occur in about 75% of people with FRDA, but they vary widely in severity. The most common abnormality in FRDA is hypertrophic cardiomyopathy, where enlarged heart muscle shrinks the blood-filled chambers in the heart, decreasing pumping capacity and leading to heart failure. It's a good idea for people with FRDA to **have regular checkups with a cardiologist.** The cardiologist will choose a treatment that is best for each patient. Regularly monitoring heart function and rhythm will allow them to treat problems early with medications or, in some instances, a pacemaker. Medications might be used to decrease the work of the heart, slow the heart rate, or decrease the chance of abnormal heart rhythm.

Certain skeletal abnormalities are also common in FRDA. Many people experience inversion (inward turning) of the feet, and a little over half have pes cavus — a shortened foot with a high arch. About two-thirds of people with FRDA develop curvature of the spine, or scoliosis.

Surgical procedures might be recommended to correct scoliosis or foot deformities that interfere with function.

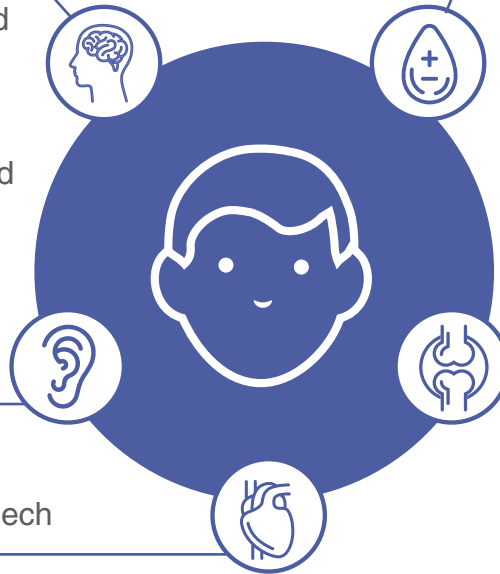
Newer **drugs that target the genetic defect in FRDA** are in development. The discovery of frataxin and its roles in iron regulation and oxidative stress have raised the possibility of treatments that might attack the underlying disease process in FRDA. Antioxidants, such as coenzyme Q10 and idebenone, have been tested in FRDA and have not shown significant benefits. However, some experts recommend them on the theory that the trials may not have been long enough or sensitive enough to show benefits.



WHAT ARE THE SIGNS AND SYMPTOMS OF FRDA?

Nervous system

- Ataxia (loss of balance and coordination)
- Dyarthria (speech that is hard to understand)
- Loss of vibration sense and position sense
- Loss of perception of temperature, light touch, and pain



Endocrine

- Prediabetes/diabetes

Hearing

- Hearing loss
- Trouble understanding speech

Heart

- Heart abnormalities
- Hypertrophic cardiomyopathy
- Arrhythmia

Skeleton and Muscle

- Scoliosis
- Weakness of the muscles of the legs and arms
- Fatigue
- Swallowing difficulties
- Muscle jerks
- Abnormal posturing of the feet or pes cavus (abnormally high arch)



To learn more about Friedreich ataxia, visit mda.org or contact the MDA Resource Center at 833-ASK-MDA1 (275-6321) or ResourceCenter@mdausa.org.

MDA GLOSSARY

Ataxia

The inability to maintain balance and coordination

Autosomal dominant spinocerebellar ataxias

A group of disorders characterized by degeneration and dysfunction of the cerebellum section of the brain and its associated pathways. These are sometimes confused with hereditary ataxias.

Autosomal recessive inheritance

One of several ways that a trait, disorder, or disease can be passed down through families. Autosomal recessive means two copies of the disease-causing gene mutation must be present for the disease or trait to develop.

Cardiac arrhythmia

Abnormal heartbeat

Diabetes

A disease in which the body's ability to produce or respond to the hormone insulin is impaired, resulting in abnormal metabolism of carbohydrates and elevated levels of glucose in the blood and urine

Hereditary ataxias

A group of neurological disorders (ataxias) that are inherited, in contrast to a related group of ataxias that are acquired through accidents, injuries, or other external agents

Hypertrophic cardiomyopathy

A condition in which part of the heart muscle becomes thickened, hindering its ability to pump blood to the body

Pes cavus

A shortened foot with a high arch

Pre-diabetes

A condition characterized by slightly elevated blood glucose levels. This is indicative that a person is at risk of progressing to Type 2 diabetes.

Reflex

An action performed as a response to a stimulus and without conscious thought

Scoliosis

An abnormal sideways curvature in the spine that occurs when weakened muscles are unable to hold the spine straight

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